CDH23 gene

cadherin related 23

Normal Function

The *CDH23* gene provides instructions for making cadherin 23, a type of protein that helps cells stick together. Different versions of this protein are made in different cell types, including a short version in the retina, which is the light-sensitive layer in the back of the eye, and a longer version in the inner ear. Cadherin 23 interacts with other proteins in the cell membrane as part of a protein complex that is involved in cell attachment.

Research suggests that the cadherin 23 protein complex helps to shape structures in the inner ear called hair bundles. These structures are made of stereocilia, which are hairlike projections that bend in response to sound waves. This bending motion is critical for converting sound waves to nerve impulses, which are then transmitted to the brain. Stereocilia are also elements of the vestibular system, the part of the inner ear that helps maintain the body's balance and orientation in space. Bending of these stereocilia is needed to transmit signals from the vestibular system to the brain.

In the retina, the role of the cadherin 23 protein complex is less well understood. Studies suggest that it plays a critical role in the function of photoreceptor cells, which are specialized cells that detect light and color.

Health Conditions Related to Genetic Changes

nonsyndromic hearing loss

Several dozen mutations in the *CDH23* gene have been identified in people with nonsyndromic hearing loss, which is loss of hearing that is not associated with other signs and symptoms. Mutations in this gene cause a form of hearing loss designated as DFNB12. This type of hearing loss begins before a child learns to speak (prelingual) and is severe to profound.

Most of the mutations that cause DFNB12 change single protein building blocks (amino acids) in cadherin 23. These mutations reduce but do not eliminate the function of this protein. The altered protein disrupts development of stereocilia in the inner ear, which leads to hearing loss.

Researchers speculate that some children with apparently nonsyndromic hearing loss caused by a *CDH23* mutation may actually have Usher syndrome (described below). A few children thought to have DFNB12 have developed the vision disorder retinitis pigmentosa later in life, which is characteristic of Usher syndrome.

Usher syndrome

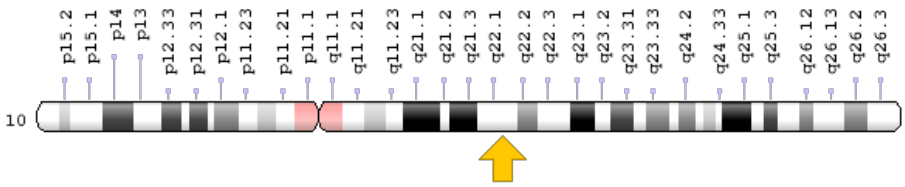
More than 60 mutations in the *CDH23* gene can cause Usher syndrome type I, which is characterized by a combination of hearing loss, vision loss, and problems with balance and coordination. Specifically, *CDH23* gene mutations cause a form of the disorder known as Usher syndrome type ID (USH1D).

Most of these mutations disrupt protein production, resulting in an abnormally small, nonfunctional version of cadherin 23 or preventing the production of any of this protein. Less frequently, mutations change single amino acids in cadherin 23. All of the *CDH23* gene mutations that cause Usher syndrome appear to eliminate the function of cadherin 23. A lack of this protein in the inner ear disrupts the normal development and function of stereocilia, which leads to hearing loss and difficulty with balance and coordination. A lack of this protein in the retina causes retinitis pigmentosa, a condition in which light-sensing cells of the retina gradually deteriorate, resulting in progressive vision loss.

Chromosomal Location

Cytogenetic Location: 10q22.1, which is the long (q) arm of chromosome 10 at position 22.1

Molecular Location: base pairs 71,396,934 to 71,815,947 on chromosome 10 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CAD23_HUMAN
- cadherin-23
- cadherin-like 23
- cadherin-related 23
- CDHR23
- DFNB12
- KIAA1774

- KIAA1812
- otocadherin
- USH1D

Additional Information & Resources

Educational Resources

- Neuroscience (second edition, 2001): Hair Cells and the Mechanoelectrical Transduction of Sound Waves
  https://www.ncbi.nlm.nih.gov/books/NBK10867/
- Neuroscience (second edition, 2001): The Retina
  https://www.ncbi.nlm.nih.gov/books/NBK10885/

GeneReviews

- Deafness and Hereditary Hearing Loss Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1434
- Usher Syndrome Type I
  https://www.ncbi.nlm.nih.gov/books/NBK1265

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CDH23%5BTIAB%5D%29+OR+%28cadherin+related+23%5BTIAB%5D%29%29+OR+%28%28cadherin-23%5BTIAB%5D%29+OR+%28otocadherin%5BTIAB%5D%29+OR+%28USH1D%5BTIAB%5D%29+OR+%28DFNB12%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- CADHERIN 23
  http://omim.org/entry/605516

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CDH23.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CDH23%5Bgene%5D
- Hereditary Hearing Loss Homepage
  http://hereditaryhearingloss.org

- HGNC Gene Family: Cadherin related
  http://www.genenames.org/cgi-bin/genefamilies/set/24
- HGNC Gene Family: Deafness associated genes
  http://www.genenames.org/cgi-bin/genefamilies/set/1152
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=13733
- Leiden Open Variation Database: CDH23 Gene Mutations
  https://research.cchmc.org/LOVD2/home.php?select_db=CDH23
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/64072
- RetNet: Summaries of Genes and Loci Causing Retinal Diseases: CDH23
  https://sph.uth.edu/retnet/disease.htm#10.201d
- UniProt
  http://www.uniprot.org/uniprot/Q9H251

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